

Survey on under-researched high-burden medical conditions

High-burden under-researched medical conditions

You are kindly invited to participate in a survey conducted by the EUHealthSupport consortium, in the context of a study supported by the European Commission's Directorate General for Research & Innovation (DG RTD).

The aim of this survey is to incorporate your opinion in an ongoing scoping study on high-burden under-researched conditions in Europe and beyond.

At this stage, we would like to capture your views on the information collected so far, and to gain more understanding what the underlying reasons might be why certain conditions are under-researched, e.g. they might have an unclear origin, an underlying mechanism that is not sufficiently understood, or may be inaccurately diagnosed.

To this end, the Commission has tasked EUHealthSupport to survey governmental funding agencies, large funding bodies and other key stakeholder groups – among others umbrella associations in the healthcare field.

Your contribution will be very much appreciated and will take approximately 15 minutes of your time. The results of this consultation will be summarised in a report, which will be submitted to the European Commission in the fall of 2022. If you have any questions about this consultation or the study itself, please contact contact@euhealthsupport.eu.

Thank you for your contribution.

About this study:

Several consecutive activities have been undertaken to come to a preliminary list of possibly under-researched and high-burden diseases and conditions: research projects related to health under the Horizon 2020 programme have been mapped with their amount of funding and the disease (group) or condition they address; this was related to the burden of these disease (groups) and conditions; a literature search identified common reasons for people to visit their general practitioner; and additional literature research provided insight into the knowledge base available for disease (groups) and conditions identified. This consultation aims to capture key stakeholders' views on how this subject is approached by others, to reflect on the list of disease (groups) and conditions that we have identified as being potentially under-researched and high-burden in past and ongoing EU Research Framework Programmes and to provide the opportunity to share your views on how this subject should be further addressed.

About us:

The EUHealthSupport Consortium is composed of Nivel (Netherlands institute for health services research; Lead), RIVM (National Institute for Public Health and the Environment of the Netherlands), Royal College of Surgeons in Ireland (RCSI), infeurope S.A., the Association of Medical Schools in Europe (AMSE) and Leginda GmbH, under the Single Framework Contract 'Chafea/2018/Health/03 - for the provision of support services for managing expert groups in the field of (public) health'. More information is available at www.euhealthsupport.eu.

Outline

This consultation starts with some background questions (part I). This will be followed by part II, in which we present our working definition on high-burden, under-researched medical conditions. Here, we ask you to reflect upon this working definition and inform us on whether and how the topic of high-burden under-researched medical conditions is currently being addressed in your organisation or country (if applicable).

Part III presents you with our preliminary shortlist of high-burden under-researched medical disease (groups) and conditions on which we would like your reflection. We would also like to hear from you what type of research would be needed to help close the research gap that exists for these disease (groups) and conditions. Finally, we ask for your thoughts about other diseases or conditions that may be under-researched while posing a high-burden on patients/society.

Part I: Background questions

Please indicate which group of stakeholders you primarily represent or consider yourself to be part of.

- EU Member State representative
- Public funding bodies (national or international)
- Philanthropic funding bodies
- Healthcare professionals
- Academia
- Industry
- Patients or citizens
- Other

Are you answering on behalf of an organisation?

- Yes
- No

Please provide the name of this organisation.

Note that all answers to this survey will be reported anonymously, but that we might consider adding a list of responding organisations as an annex to the final report.

World ME Alliance

What is your (organisation's) disease area of expertise, if any?

Myalgic Encephalomyelitis/Chronic Fatigue Syndrome (ME/CFS)

Please indicate at which geographical level your organisation is predominantly active.

- Local or regional level within one country
- National level within one country
- European level (covering multiple European countries)
- International level (other or wider than Europe)

In which country is your organisation based?

United Kingdom

Part II: Views on working definition

In this study, we defined high-burden under-researched diseases or conditions as *those diseases or conditions that receive insufficient research funding relative to the expected level of funding based on their burden.*

With this definition, we aim to identify those medical conditions where the research funding is insufficient to close the gap in robust scientific evidence needed for improved policies and practices to tackle these medical conditions. This can result in a lack of knowledge that hampers adequate preventive, diagnostic and therapeutic strategies, while the disease poses a considerable burden on patients in terms of incidence /prevalence, mortality or quality of life.

What is your reflection on our working definition?

In case your organisation or country has its own definition of high-burden under-researched medical conditions, or in case you have suggestions for another definition or an addition to the above mentioned definition, please mention this here as well.

We are concerned that the working definition fails to identify a group of underresearched high-burden diseases, but could be argued subjectively to relate to many diseases.

The lack of a definition of "expected level of funding" means that some well-funded diseases could still become part of the new category of medical conditions you aim to identify. We also think that the size of the "gap in robust scientific evidence" that is mentioned in your explanation of the definition is important and should be included as an explicit criterion.

We therefore recommend adopting a stricter definition, that accounts for the impact that a lack of treatment options and understanding of disease etiology can have on funding. Both the absence of a therapy and a poorly understood etiology are hallmarks of a gap in scientific evidence that needs to be addressed in this explicit criterion.

While low "funding per burden" is an important criterion for a disease to be neglected, there are also further factors. Many of the diseases that should be included in this new category have so little research funding or research infrastructure that they are yet to be adequately recognised by international bodies, let alone addressed. It is these diseases, in particular, that need adaptations of the current frameworks to be compatible for funding. Examples include ME/CFS, Postural Orthostatic Tachycardia Syndrome (POTS), hypermobile Ehlers Danlos Syndrome (hEDS), and more.

Many diseases falling into this grouping do not have a strong evidence base for the burden they cause, yet the research that is available demonstrates a devastating impact on quality of life, for the individual and their families/carers. We want clear language that assures that the lack of research and recognition does not further exclude these diseases from being a named part of a disease grouping of "under-researched" diseases.

If you are aware of (national) research or R&I initiatives that have been undertaken to address underresearched conditions in Europe over the last 5 years, please let us know here.

If relevant, please provide links to any public document that describes or explains how underresearched conditions are defined or addressed in that initiative

The UK ME/CFS Priority Setting Partnership was established to identify the top priorities for research into ME /CFS, "which are of direct relevance and potential benefit to patients and the clinicians who treat them". Results available here: <https://psp-me.co.uk/>

The UK Department of Health and Social Care announced a delivery plan on ME/CFS, which includes a Research Working Group tasked with developing a research strategy for ME/CFS in the UK, building capacity and capability, and raising awareness and building trust. This process is ongoing, information about it will continue to be released through various charity websites such as actionforme.org.uk

Considering the lack of available evidence in the EU, we consider it important to take note of research elsewhere. A study undertaken in the US found that "ME/CFS [research was] receiving roughly 7% of that commensurate with disease burden." <https://pubmed.ncbi.nlm.nih.gov/32568148/>

Understanding the current situation is always a first step toward creating change.

Part III: Views on high-burden under-researched (groups of) diseases

In this study, several consecutive activities have been undertaken to come to a preliminary list of possibly under-researched and high-burden diseases and conditions: research projects related to health under the Horizon 2020 programme have been mapped with their amount of funding and the disease (group) or condition they address; this was related to the burden of these disease (groups) and conditions; a literature search identified common reasons for people to visit their general practitioner; and additional literature research provided insight into the knowledge base available for disease (groups) and conditions identified. This has resulted in a preliminary overview of possibly under-researched disease (groups) and conditions that are high-burden for patients in the EU (shown below). What we would like to know from you is: a) Do you recognise these disease (groups) and conditions as being underresearched? b) What type of research do you think is especially needed? and c) What other high-burden diseases may be underresearched that are not included in the list?

The following list of disease (groups) and conditions have been identified as possible underresearched and high-burden to patients in Europe:

(please note that the research underlying the creation of this list has its limitations; this list is debatable and therefore your expert opinion is needed)

- Mental disorders
 - Mental disorders
 - Depression or anxiety
 - Self-harm
- Disorders of the blood and organ system
 - Chronic kidney disease
 - Cirrhosis and other chronic liver diseases
 - Diabetes mellitus
 - Cystitis / urinary tract infection
 - Gallbladder and biliary diseases
 - Dyspepsia
 - Abdominal pain
 - Hemoglobinopathies and hemolytic anemias
- Musculoskeletal disorders
 - Musculoskeletal disorders
 - Arthritis (not back)
 - Injuries
 - Low back pain
- Headache disorders
 - Headache disorders
 - Tension-type headaches
 - Migraine
- Fatigue / weakness
- Sleep-wake disorders
- Skin and subcutaneous diseases
 - Dermatitis
- Sense organ diseases
- Gynecological diseases
 - Pregnancy, childbirth or the puerperium

- Conditions related to sexual health

Do you recognise the disease (groups) and/or conditions in the above list as being under-researched?

- Yes, all of the disease groups on the list
- Yes, part of the disease groups on the list
- No, none of the disease groups on the list
- I don't know

Please specify which disease (groups) and/or conditions should not be included in the list:

You will be given the opportunity to suggest disease (groups) and/or conditions that should be included but are not part of the above list later on.

We do not believe that any larger disease "groups" can or should be included on this list, as there will always be more and less researched diseases within any group. We therefore believe all groupings and symptoms should be removed - instead there needs to be a clear list of underfunded diseases so that research funding that we hope will be allocated to this category genuinely goes to underfunded high-burden diseases.

That said, we would particularly note that diabetes mellitus, arthritis, depression, kidney disease, and others have major research networks, hundreds of millions of euros in funding, and seem surprising additions when diseases such as ME/CFS, fibromyalgia and endometriosis are not even named.

Please select a maximum of three (groups of) diseases / conditions from the list below on which you consider yourself to be an expert.

- Mental disorders (including, but not limited to: depression or anxiety, and self-harm)
- Disorders of the blood and organ system (including, but not limited to: chronic kidney disease, cirrhosis and other chronic liver diseases, diabetes mellitus, cystitis / urinary tract infection, gallbladder and biliary diseases, dyspepsia, abdominal pain, and hemoglobinopathies and hemolytic anemias)
- Musculoskeletal disorders (including, but not limited to: arthritis (not back), injuries, and low back pain)
- Headache disorders (including, but not limited to: tension-type headaches, and migraine)
- Fatigue / weakness
- Sleep-wake disorders
- Skin and subcutaneous diseases (including, but not limited to: dermatitis)
- Sense organ diseases
- Gynecological diseases (including, but not limited to: pregnancy, childbirth or the puerperium)
- Conditions related to sexual health

What are in your opinion the minimal research requirements needed for this type of research, to make a real progress/impact?

- Cohorts
- Biobanks
- Large databases like genetic/imaging data
- Clinical trial networks
- Other

Fatigue / weakness

Can you indicate whether you think this is indeed an under-researched (group of) disease(s) with a high burden for patients?

- I am certain that this is an under-researched high burden (group of) disease(s)
- I suspect that this may be an under-researched high burden (group of) disease(s)
- I am not sure that this group is an under-researched high burden (group of) disease(s)
- I am certain that this group is NOT an under-researched high burden (group of) disease(s)

What type of research would be primarily needed to close the evidence gap allowing for improved policies and practices to tackle these medical conditions?

- Prevention
- Basic or fundamental research
- Clinical (incl. diagnostic and/or treatment)
- Implementation
- Other
- I don't know

What are in your opinion the minimal research requirements needed for this type of research, to make a real progress/impact?

- Cohorts
- Biobanks
- Large databases like genetic/imaging data
- Clinical trial networks
- Other

What do you think should be improved in order to increase the research activities?

- Create more / better national, EU, and international funding opportunities
- Increase incentives (other than financial) to do research on these medical conditions
- Make incentives sufficiently accessible to do research on these medical conditions
- Stimulate the organisation of specific conferences in the medical areas
- Stimulate researcher mobility
- Stimulate the creation of large research networks
- Other

Please specify how you would envision incentives can be made sufficiently accessible

Can you indicate other conditions, diseases or disease groups that you would consider as 'under-researched' and high burden to patients, in need of robust scientific evidence for improved management of these medical conditions?

If relevant, please provide links to any public document that describes or explains how under-researched conditions are defined or addressed in your country. Please be aware that rare diseases are not within the scope of this study.

- No
- Yes

Please specify:

Myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS), postural tachycardia syndrome (POTS), hypermobile Ehlers Danlos Syndrome (EDS), Chronic Lyme disease, fibromyalgia, endometriosis, mast cell activation syndrome (MCAS).

Can you think of any (European) Research Networks on these conditions?

- No
 Yes

Can you think of ways to enhance the research capacity in Europe for these conditions?

- No
 Yes

Please exemplify for all the disease (groups) and/or conditions that you have specified:

We wish to see funding opportunities that are specific enough to these conditions that other areas with greater existing research capacity do not out compete them. We believe clear funding pots and public commitments to address these diseases would garner interest and lead to new research that we hope would snowball to even greater capacity in Europe and worldwide.

Can you think of any possible reasons explaining why these medical conditions are understudied and underrepresented in EU-funded projects?

- No
 Yes

Please exemplify for all the disease (groups) and/or conditions that you have specified:

We see three key reasons for the fact that these diseases remain understudied:

1. Lack of understanding of etiology
2. Stigma and lack of awareness of disease impact
3. Bias against diseases predominantly affecting women.

Without changing approaches to research funding, these are likely to remain the case.

Many of the diseases we have mentioned (ME/CFS, fibromyalgia etc.) are diagnosed via their clinical symptoms, but have no established diagnostic biomarker. As a consequence, many patients remain undiagnosed and there remains significant stigma attached to each.

A funding scheme prioritising neglected diseases with a high burden could help address this, not just through understanding disease etiology but by creating and reinforcing an entire ecosystem of research that helps to build centres of excellence, create pathways for patients to become involved in research, and change public perceptions of the diseases. Each of these in turn would then accelerate research further, but without an initial funding effort to jump-start the process, research in these areas is bound to remain severely lacking.

Are there in your opinion any disease (groups) and/or conditions with significant knowledge gaps related to specific population group(s)?

E.g. for women, elderly, persons with a migration background, socially vulnerable persons, etc.

Many high-burden multi-system diseases such as ME/CFS affect more women than men, potentially due to a role of auto-immunity. Research suggests that gender bias impacts research spending. We also believe that ME/CFS is under-diagnosed in ethnic minority populations.

Finally we find that many people with ME/CFS live with multiple comorbidities. Multi-morbidity is associated with poor health outcomes, and in itself an underresearched area.

Are there any final comments that you would like to make?

We believe that the establishment of a new category of neglected high-burden diseases, through this scoping study, could be vital to invigorating research into some of the most neglected diseases to date, which affect millions of patients in the EU and world-wide. However, this requires a well-designed definition that focuses on severe underfunding, lack of disease knowledge and therapeutic options. The list of diseases and groups included so far appears to instead re-highlight diseases that are commonly part of other widely accepted and used categories and have established routes to gain funding.

We would like to see greater specificity throughout, and for this project to consider the impact of low levels of research to date on the diseases they are assessing for inclusion.

We are excited by the potential of this new category to create system change in the way neglected high-burden diseases are approached, and hope this will not be a wasted opportunity.

Thank you for your cooperation.

GETTING IN TOUCH WITH THE EU

In person

All over the European Union there are hundreds of Europe Direct information centres. You can find the address of the centre nearest you at:

https://europa.eu/european-union/contact_en

On the phone or by email

Europe Direct is a service that answers your questions about the European Union. You can contact this service:

– by freephone: 00 800 6 7 8 9 10 11 (certain operators may charge for these calls),

– at the following standard number: +32 22999696, or

– by email via: https://europa.eu/european-union/contact_en

FINDING INFORMATION ABOUT THE EU

Online

Information about the European Union in all the official languages of the EU is available on the Europa website at: https://europa.eu/european-union/index_en

EU publications

You can download or order free and priced EU publications from: <https://op.europa.eu/en/publications>. Multiple copies of free publications may be obtained by contacting Europe Direct or your local information centre (see https://europa.eu/european-union/contact_en).

EU law and related documents

For access to legal information from the EU, including all EU law since 1952 in all the official language versions, go to EUR-Lex at: <http://eur-lex.europa.eu>

Open data from the EU

The EU Open Data Portal (<http://data.europa.eu/euodp/en>) provides access to datasets from the EU. Data can be downloaded and reused for free, for both commercial and non-commercial purposes.



Contact

contact@euhealthsupport.eu